

# 17<sup>th</sup> UK Neuromuscular Translational Research Conference

## ***Developing Advanced Therapies in Neuromuscular Diseases***

Goodenough College Mecklenburgh Square, London WC1N 2AB

17<sup>th</sup> and 18<sup>th</sup> April 2024

### Day 1 – Wednesday 17th April

09:00 – 09:30            **Registration and Coffee**

09:30 – 09:45            **Welcome and Introduction**  
Professor Michael Hanna  
Director, ICGNMD and UCL Institute of Neurology

### Session 1: Latest Developments in Genetic Therapies in NMD - challenges and opportunities

Chairs: Professor Volker Straub (Newcastle University), Professor Mary Reilly (UCL IoN)

09:45 – 10:30            **Safety of Gene Therapies**  
Professor Francesco Muntoni  
UCL GOS Institute of Child Health and Queen Square Institute of Neurology  
Plenary Lecture

10:30 – 11:00            **Accelerating new therapies for rare neuromuscular diseases**  
Professor Matthew Wood  
MDUK Oxford Neuromuscular Centre

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11:00 – 11:30            **Coffee**

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### Platform presentations

11:30 – 12:45            Platform presentation 1  
**DOK7-AAV9 gene therapy in a novel mouse model for Congenital Myasthenic Syndrome caused by mutations in CHRND**  
Dr. Setareh Alabaf  
University of Oxford

Platform presentation 2  
**Antisense oligonucleotide mediated mutant Sptlc1 silencing as a therapeutic strategy for hereditary sensory neuropathy type IA: A proof of concept study in a mouse model**  
Dr. Jinhong Meng  
UCL GOS Institute of Child Health

Platform presentation 3  
**Differential TfR1 expression patterns impact TfR1 targeting delivery strategies in neuromuscular disease models**  
Jessica Stoodley

University of Oxford

Platform presentation 4

**Therapeutic effect of AAV-mediated delivery of linker proteins in a mouse model for LAMA2-related congenital muscular dystrophy**

Dr. Judith Reinhard  
University of Basel

Platform presentation 5

**Efficacy and Safety of anti FcRN treatment in Refractory MG - the UK experience of the Efgartigimod Early Access to Medicine**

Dr. Jen Spillane  
National Hospital for Neurology and Neurosurgery, Queen Square

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12:45 – 13:00      **MDUK Update**  
Cat Woodhead  
CEO  
Muscular Dystrophy UK

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13:00 – 13:45      **Lunch**

13:45 – 14:45      Guided poster sessions

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**Session 1 (ctd): Latest Developments in Genetic Therapies in NMD - challenges and opportunities**

Chairs: Professor Bobby McFarland (Newcastle University) and Professor Rita Horvath (University of Cambridge)

14:45 – 15:15      **Treatments for TTR Amyloidosis**  
Professor Julian Gillmore  
UCL Centre for Amyloidosis

15:15 – 15:45      **DMD / DM1 Therapy Advances - Next Generation RNA Therapies Update**  
Dr. Aurélie Goyenvalle  
Director Biotherapy for Neuromuscular Diseases, University of Versailles, INSERM

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15:45 – 16:15      **Coffee**

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**Platform presentations**

Chairs: Professor Bobby McFarland (Newcastle University) and Professor Rita Horvath (University of Cambridge)

16:15– 17:00      Platform presentation 6  
**In vitro 3D-model of mitochondrial myopathy human skeletal muscle**  
Dr. Valeria Di Leo  
Wellcome Centre for Mitochondrial Research, Newcastle University

Platform presentation 7

**Defective Mitochondria Induce Quantitative Easing of Cholesterol Leading to Neurological Disease**

Professor Antonella Spinazzola  
UCL Queen Square Institute of Neurology

Platform presentation 8

**Differences in executive function skills used during planning and navigation observed in boys with DMD**

17:00 – 17:10      **Break**

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## Session 2: Applications of Exercise Therapy in the era of Gene Therapy in NMD

Chairs: Professor Francesco Muntoni (UCL GOSH ICH) and Dr Anna Sarkozy (Great Ormond Street Hospital)

17:10 – 18:00      **The Sixth Victor Dubowitz Lecture**  
Introduced by Prof Francesco Muntoni

### **Can all persons with neuromuscular diseases benefit from exercise training?**

Professor John Vissing  
Director, Copenhagen Neuromuscular Centre  
Rigshospitalet, Copenhagen

18:00 – 18:20      **MRC – UKRI update**  
Introduced by Professor Michael Hanna  
Professor Patrick Chinnery  
Executive Chair, Medical Research Council

18:30                **Networking reception and canapés**  
Goodenough College, Mecklenburgh Square, London WC1N 2AB

## Day 2 – Thursday 18th April

08:15 Coffee

08:30 - 08:45 **Welcome**

### Session 2 (ctd): Applications of Exercise Therapy in the era of Gene Therapy in NMD

Chairs: Professor Francesco Muntoni (UCL GOSH ICH) and Dr. Anna Sarkozy (Great Ormond Street Hospital)

08:45 – 09:30 **Role of Physiotherapy Rehabilitation in the Landscape of Genetic Therapies**

Dr. Gita Ramdharry

Queen Square Centre for Neuromuscular Diseases

National Hospital for Neurology & Neurosurgery, UCLH

UCL Queen Square Institute of Neurology

### Session 3: Mechanisms and New Advanced Therapeutics in Autoimmune Neuromuscular Disease

Chairs: Professor Michael Hanna (UCL IoN) and Professor Rob Pitceathly (UCL IoN)

09:30– 10:15 **Innate immune system induction in mitochondrial disease**

Professor Yanick Crow

MRC Human Genetics Unit, University of Edinburgh

10:15 – 10:45 **Immunological Disease Mechanisms and New Treatments in Inflammatory Myopathies**

Professor Pedro Machado

UCL Institute of Neurology

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10:45 – 11:15 **Break / Coffee**

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### Poster flash sessions

11:15 – 12:15 **Raman spectroscopy as a translational biomarker of muscle health**

Dr. James Alix

Sheffield Institute for Translational Neuroscience

**A remarkable response to a new treatment in a patient with refractory myasthenia gravis: a case report with video recordings**

Dr. Mohammed Ashraghi

University of Oxford

**Advanced in vitro modelling of LMNA-related congenital muscular dystrophies using patient-derived iPSCs for development of gene editing strategies**

Daniel Moore

University College London

**Investigating mitochondrial alanyl-tRNA synthetase defects in neurons**

Oliver Podmanicky

Dept of Clinical Neurosciences, University of Cambridge

**Investigating loss of Replication Factor Complex subunit 1 (RFC1) function in CANVAS patients and heterozygous AAGGG expansion carriers**

Dr. Ricardo Schnekenberg

UCL Institute of Neurology

**Using innovative Data Modelling methods to improve data quality- learning from Adult SMA REACH a real-world data collection study**

Sonia Segovia Simon

John Walton Muscular Dystrophy Research Centre, Newcastle University

**Interactome analysis of dystrophin isoforms in the mouse brain**

Dr. Konstantina Teterou

Dubowitz Neuromuscular Centre, Institute of Child Health, UCL

**Retrospective deep phenotypic and genotypic analysis of UK patients with recessive early onset titinopathy**

Dr. SiewMei Yap

Department of Neurology, Royal Free Hospital

**Targeting adipogenic differentiation of fibro-adipogenic precursors in Duchenne muscular dystrophy**

Priyanka Mehra

John Walton Muscular Dystrophy Research Centre, Newcastle University

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**Platform presentations**

Chairs: Professor Michael Hanna (UCL IoN) and Professor Rob Pitceathly (UCL IoN)

12:15 – 12:45

Platform presentation 9

**Uplift in diagnosis after reanalysis of WES data in a 10-year-old cohort of patients with suspected mitochondrial disorders**

Dr. Catarina Olimpio

Department of Clinical Neurosciences, University of Cambridge

Platform presentation 10

**Optimising a muscle-specific AAV-BDNF gene therapy in mouse models of Charcot-Marie-Tooth Disease**

Dr. Ellie Rhymes

UCL Queen Square Institute of Neurology

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12:45 – 13:30

**Lunch**

13:30 – 14:30

Guided poster sessions

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**Session 3 (ctd): Mechanisms and New Advanced Therapeutics in Autoimmune Neuromuscular Disease**

Chairs: Professor Mary Reilly (UCL IoN) and Dr. Alex Rossor (UCL IoN)

14:30 – 15:00

**Novel Treatments for Immune Mediated Neuropathies including CIDP, Nodopathies and GBS**

Professor Mike Lunn

UCL Queen Square Institute of Neurology

15:00 – 15:30

**Autoimmune Myasthenic Syndrome - The Recent Dramatic Progress in Treatment of Autoimmune Myasthenia Gravis**

Dr. Jennifer Spillane

Consultant Neurologist

UCLH NHNN

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15:30 – 16:00

**Break / Coffee**

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## Session 4: New Genomic Advances in Neuromuscular Disease

Chairs: Professor Rob Taylor (Newcastle University) and Professor Henry Houlden (UCL IoN)

16:00 – 16:35

**The Sixth Morgan-Hughes Thomas lecture**  
**Introduced by Professor Henry Houlden (UCL IoN)**

**Improved diagnosis of patients through systematic variant resolution**

Professor Conrad (Chris) Weihl  
Head of Weihl Lab, University of Washington in St Louis

16:35 – 17:05

**Gene Modifiers in Mitochondrial Diseases**

Dr. Sarah Pickett  
Wellcome Trust Centre for Mitochondrial Research, Newcastle University

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17:00 – 17:15

**Prizes and thanks**

Professor Mike Hanna and Professor Mary Reilly

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